PRENATAL DIAGNOSIS
(Prenat. diagn.)

CONTENTS

VOLUME 14, ISSUE Nos. 1–12, PLUS ANNUAL REVIEW ISSUE

ISSUE NUMBER 1, JANUARY 1994

Review Article

Recent Advances in the Prenatal Diagnosis of the Mucopolysaccharidoses: A. H. FENSOM and P. F. BENSON 1

Original Papers

Cytogenetic Abnormalities Among Perinatal Deaths Demonstrating a Single Umbilical Artery: D. N. SALLER Jr. and R. NEIGER 13

Prenatal Diagnosis of Sanfilippo Disease Type C Using a Simple Fluorometric Enzyme Assay: W. HE, Y. V. VOZNYI, J. G. M. HUIJMANS, G. C. GEILEN, E. A. KARPOVA, T. V. DUDUKINA, J. ZAREMBA, O. P. VAN DIGGELEN and W. J. KLEIJER 17

Prenatal Identification of an Isochromosome for the Short Arm of the Y i(Yp), by Cytogenetic and Molecular Analyses: R. SLIM, J. SOULIÉ, J. HOTMAR, B. LECOLIER, G. BERCAU and A. BERNHEIM 23

Methylenetetrahydrofolate Reductase Deficiency: Prenatal Diagnosis and Family Studies: J. MARQUET, B. CHADEFAUX, J. P. BONNEFONT, J. M. SAUDUBRAY and J. ZITTOUN 29

Implications of Maternal Serum Alpha-Fetoprotein Elevation Caused by Transabdominal and Transcervical CVS: S. SMIDT-JENSEN, J. PHILIP, J. M. ZACHARY, S. E. FOWLER and B. NØRGAARD-PEDERSEN 35

Short Communications

Sonographic Findings with Pfeiffer Syndrome: L. M. HILL and P. C. GRZYBEK 47

First-Trimester Prenatal Diagnosis in Twin Pregnancies: G. C. M. L. CHRISTIAENS, J. C. OOSTERWIJK, R. H. STIGTER, P. P. DEUTZ-TERLOUW, A. L. J. KNEPPERS and E. BAKKER 51

Prenatal Diagnosis of X-Linked Hydrocephalus in a Chinese Family with Four Successive Affected Pregnancies: T-M. KO, H-L. HWA, L-H. TSENG, F-J. HSIEH, S-F. HUANG and T.-Y. Lee 57

Hereditary Multiple Intestinal Atresia—Ultrasound Findings and Outcome of Pregnancy in an Affected Case: P. A. BOYD, P. CHAMBERLAIN, S. GOULD. N. K. IVES, N. MANNING and T. TSANG 61

Human Chorionic Gonadotropin and Unconjugated Oestriol Measurements in Insulin-Dependent Diabetic Pregnant Women Being Screened for Fetal Down Syndrome: G. E. PALOMAKI, G. J. KNIGHT and J. E. HADDOW 65

Letters to the Editor

On a Case of Fryns' Syndrome: A. DI MEGLIO, G. DI MEGLIO, A. RICCIO and V. B. FERRIGNO 69

Is Non-Echogenic Nuchal Oedema A Marker in Trisomy 21 Screening?: R. SAURA, J. L. BRUN, L. TAINE, J. HOROVITZ,. B. MAUGEY and D. ROUX 69

Operator Experience and Fetal Loss Rate in Transabdominal CVS: R. SAURA, B. GAUTHIER, L. TAINE, Z. Q. WEN, J. HOROVITZ, D. ROUX, B. LAULOM and A. VERGNAUD 70

Brain Echography in Newborns to Determine Whether Chorionic Villus Sampling Causes Lesions: C. FABRIS, D. LICATA, C. MARTANO, M. MOMBRO, R. BAGNA and M. MOSTERT 71

Prenatal Diagnosis of a Heterochromatic 18p+ Heteromorphism: M. C. PITTALIS, L. SANTARINI and L. BOVICELLI 72

Prenatal Diagnosis of Thrombocytopenia in TAR Syndrome: A. E. DONNENFELD .. 73

Prenatal Diagnosis of Thrombocytopenia in TAR Syndrome: M. G. BIALER 74

PRENATAL DIAGNOSIS
(Prenat. diagn.)

CONTENTS

VOLUME 14, ISSUE Nos. 1–12, PLUS ANNUAL REVIEW ISSUE

ISSUE NUMBER 1, JANUARY 1994

Review Article

Recent Advances in the Prenatal Diagnosis of the Mucopolysaccharidoses: A. H. FENSOM and P. F. BENSON 1

Original Papers

Cytogenetic Abnormalities Among Perinatal Deaths Demonstrating a Single Umbilical Artery: D. N. SALLER Jr. and R. NEIGER 13

Prenatal Diagnosis of Sanfilippo Disease Type C Using a Simple Fluorometric Enzyme Assay: W. HE, Y. V. VOZNYI, J. G. M. HUIJMANS, G. C. GEILEN, E. A. KARPOVA, T. V. DUDUKINA, J. ZAREMBA, O. P. VAN DIGGELEN and W. J. KLEIJER 17

Prenatal Identification of an Isochromosome for the Short Arm of the Y i(Yp), by Cytogenetic and Molecular Analyses: R. SLIM, J. SOULIÉ, J. HOTMAR, B. LECOLIER, G. BERCAU and A. BERNHEIM 23

Methylenetetrahydrofolate Reductase Deficiency: Prenatal Diagnosis and Family Studies: J. MARQUET, B. CHADEFAUX, J. P. BONNEFONT, J. M. SAUDUBRAY and J. ZITTOUN 29

Implications of Maternal Serum Alpha-Fetoprotein Elevation Caused by Transabdominal and Transcervical CVS: S. SMIDT-JENSEN, J. PHILIP, J. M. ZACHARY, S. E. FOWLER and B. NØRGAARD-PEDERSEN 35

Short Communications

Sonographic Findings with Pfeiffer Syndrome: L. M. HILL and P. C. GRZYBEK 47

First-Trimester Prenatal Diagnosis in Twin Pregnancies: G. C. M. L. CHRISTIAENS, J. C. OOSTERWIJK, R. H. STIGTER, P. P. DEUTZ-TERLOUW, A. L. J. KNEPPERS and E. BAKKER 51

Prenatal Diagnosis of X-Linked Hydrocephalus in a Chinese Family with Four Successive Affected Pregnancies: T-M. KO, H-L. HWA, L-H. TSENG, F-J. HSIEH, S-F. HUANG and T.-Y. Lee 57

Hereditary Multiple Intestinal Atresia—Ultrasound Findings and Outcome of Pregnancy in an Affected Case: P. A. BOYD, P. CHAMBERLAIN, S. GOULD. N. K. IVES, N. MANNING and T. TSANG 61

Human Chorionic Gonadotropin and Unconjugated Oestriol Measurements in Insulin-Dependent Diabetic Pregnant Women Being Screened for Fetal Down Syndrome: G. E. PALOMAKI, G. J. KNIGHT and J. E. HADDOW 65

Letters to the Editor

On a Case of Fryns' Syndrome: A. DI MEGLIO, G. DI MEGLIO, A. RICCIO and V. B. FERRIGNO 69

Is Non-Echogenic Nuchal Oedema A Marker in Trisomy 21 Screening?: R. SAURA, J. L. BRUN, L. TAINE, J. HOROVITZ,. B. MAUGEY and D. ROUX 69

Operator Experience and Fetal Loss Rate in Transabdominal CVS: R. SAURA, B. GAUTHIER, L. TAINE, Z. Q. WEN, J. HOROVITZ, D. ROUX, B. LAULOM and A. VERGNAUD 70

Brain Echography in Newborns to Determine Whether Chorionic Villus Sampling Causes Lesions: C. FABRIS, D. LICATA, C. MARTANO, M. MOMBRO, R. BAGNA and M. MOSTERT 71

Prenatal Diagnosis of a Heterochromatic 18p+ Heteromorphism: M. C. PITTALIS, L. SANTARINI and L. BOVICELLI 72

Prenatal Diagnosis of Thrombocytopenia in TAR Syndrome: A. E. DONNENFELD .. 73

Prenatal Diagnosis of Thrombocytopenia in TAR Syndrome: M. G. BIALER 74

Absence or Reduction of total IgM in Fetal Blood as an Indication for Cytogenetic Analysis?: P. HOHLFELD, Y. VIAL and G. PESCIA 75

ISSUE NUMBER 2, FEBRUARY 1994

Evaluation of X, Y, 18, and 13/21 Alpha Satellite DNA Probes for Interphase Cytogenetic Analysis of Uncultured Amniocytes by Fluorescence *in Situ* Hybridzation: V. CACHEUX, G. TACHDJIAN, L. DRUART, J. F. OURY, S. SÉRERO, P. BLOT, and C. NESSMANN 79

Fluorescence *in situ* Hybridization with a Chromosome 21-Specific Cosmid Contig: 1-Day Detection of Trisomy 21 in Uncultured Mesenchymal Chorionic Villus Cells: T. BRYNDORF, B. CHRISTENSEN, Y. XIANG, J. PHILIP, K. YOKOBATA, N. BUI and C. GAISER 87

Maternal Serum Free Beta hCG Screening: Results of Studies Including 480 Cases of Down Syndrome: J. N. MACRI, K. SPENDER, K. GARVER, P. D. BUCHANAN, B. SAY, N. J. CARPENTER, F. MULLER and A BOUÉ 97

Tertiary Centre Referral of Small-for-Gestational Age Pregnancies: A 10-year Retrospective Analysis: R. HEYDANUS, I. P. VAN SPLUNDER and J. W. WLADIMIROFF 105

Maternal Contamination of Amniotic Fluid Demonstrated by DNA Analysis: M. T. REBELLO, A. ABAS, K. NICOLAIDES and D. V. COLEMAN 109

Six Years' Experience with Rapid Karyotyping in Prenatal Diagnosis: Correlations Between Phenotype Detected by Ultrasound and Fetal Karyotype: U. CLAUSSEN, R. ULMER, E. BEINDER and H-J. VOIGT 113

Carrier Detection and Microsatellite Analysis of Duchenne and Becker Muscular Dystrophy in Spanish Families: H. KRUYER, M. MIRANDA, V. VOLPINI and X. ESTIVILL 123

Screening of Monoclonal Antibodies Recognizing Oncofetal Antigens for Isolation of Trophoblasts from Maternal Blood for Prenatal Diagnosis: L. G. DURRANT, K. M. McDOWELL, R. A. HOLMES and D. T. Y. LIU 131

Short Communications

Meckel-Gruber Syndrome: Prenatal Diagnosis at 10 Menstrual Weeks Using Embryoscopy: Y. DUMEZ, M. DOMMERGUES, M-C. GUBLER, V. BUNDUKI, F. NARCY, M. LeMERRER, L. MANDELBROT and R. BERKOWITZ 141

Prenatal Exclusion of Stickler Syndrome: J. ZLOTOGORA, M. GRANAT and R. G. KNOWLTON 145

Early Prenatal Direct Gene Diagnosis of Cystic Fibrosis in Twin Pregnancy and Subsequent Selective Termination: F. S. JØRGENSEN, J. BANG, L. TRANEBJAERG, L. N. BERGE, S. H. EIK-NES and M. SCHWARTZ 149

Letter to the Editor

Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome: A Case Report Supporting Autosomal Recessive Inheritance: H. M. McNAMARA, J. L. ONWUDE and J. G. THORNTON 153

ISSUE NUMBER 3, MARCH 1994

Original Papers

Transabdominal Chorionic Villus Sampling and Amniocentesis for Prenatal Diagnosis: 5 Years' Experience at a University Centre: P. PALO, O. PIIROINEN, E. HONKONEN, T. LAKKALA and P. AULA 157

Mosaic Triple Trisomy in Amniocytes from a Phenotypically and Karyotypically Normal Fetus: J. T. MASCARELLO, M. C. JONES, V. A. CATANZARITE and K. H. BROWN 163

Tibial Hemimelia Syndrome: Prenatal Diagnosis by Real-time Ultrasound: M. RAMIREZ, J. T. HECHT, S. TAYLOR and I. WILKINS 167

A Fetus with a Parvovirus B19 Infection and Congenital Anomalies: R. G. TIESSEN, A. M. W. VAN ELSACKER-NIELE, CHR. VERMEIJ-KEERS, D. OEPKES, J. VAN ROOSMALEN and M. C. B. GORSIRA 173

The Importance of Late Pregnancy Scans for Renal Tract Abnormalities: G. ECONOMOU, J. A. EGGINTON and D. S. K. BROOKFIELD 177

Human Parvovirus B19 Infection and Unbalanced Translocation in a Case of Hydrops Fetalis: C. WILLEKES, F. J. M. E. ROUMEN, A.-M. W. VAN ELSACKER-NIELE, H. T. WEILAND, C. VERMEY-KEERS, J. H. J. M. VAN KREIKEN, C. E. M. DE DIE-SMULDERS, G. J. H. HAMERS and G. P. M. VAES-PETERS 181

Prenatal Diagnosis of Congenital Diaphragmatic Hernia: A Retrospective Analysis of 28 Cases: M. MANNI, R. HEYDANUS, N. S. DEN HOLLANDER, P. A. STEWART, CH. DE VOGELAERE and J. W. WLADIMIROFF 187

Fetal Diagnosis of Toxoplasmosis in 190 Women Infected During Pregnancy: F. PRATLONG, P. BOULOT, E. ISSERT, M. MSIKA, F. DUPONT, B. BACHELARD, P. SARDA, J.-L. VIALA and D. JARRY 191

The Influence of Serum Screening on the Amniocentesis Rate in Women of Advanced Maternal Age: J. R. BEEKHUIS, B. T. H. M. DE WOLF, A. MANTINGH and M.P. HERINGA 199

Maternal Serum Human Chorionic Gonadotrophin and Pregnancy-Associated Plasma Protein A, Markers for Fetal Down Syndrome at 8–14 weeks: M. C. M. MacINTOSH, R. ILES, B. TEISNER, K. SHARMA, T. CHARD, J. G. GRUDZINSKAS, R. H. T. WARD and F. MULLER 203

Early Genetic Amniocentesis and its Relationship to Respiratory Difficulties in Paediatric Patients: A Report of Findings in Patients and Matched Controls 3–5 Years Post-Procedure: B. C. CALHOUN, W. BREHM and A. T. BOMBARD 209

Re-evaluation of the Colorimetric Assay for Cytidine Deaminase Activity: T. OKAMURA and K. KIGASAWA 213

Short Communications

Prenatal Diagnosis of Werdnig-Hoffman Disease: DNA Analysis of a Mummified Umbilical Cord Using Closely Linked Microsatellite Markers: T. MATILLA, J. CORRAL, M. MIRANDA, J. TROYANO, K. MORRISON, V. VOLPINI and X. ESTIVILL .. 219

Significance of Septations in Isolated Fetal Cystic Hygroma Detected in the First Trimester: L. P. SHULMAN, N. A. RAAFAT, P. C. MACE, D. S. EMERSON, R. E. FELKER, J. L. SIMPSON and S. ELIAS 223

Prenatal Detection of X-Linked Ichthyosis by Maternal Serum Screening for Down Syndrome: I. BARTELS, J. CAESAR and U. SANCKEN 227

Annotation

A Simple Alternative to Amniocentesis?: M. ADINOLFI, P. SOOTHILL and C. RODECK 231

ISSUE NUMBER 4, APRIL 1994

Original Papers

Prenatal Exclusion of Duchenne Muscular Dystrophy by Fetal Muscle Biopsy: R. J. BENZIE, P. RAY, D. THOMPSON, A. G. W. HUNTER, B. IVEY and L. SALVADOR 235

Abnormal Immunological Development in Fetuses with Trisomy 18: G. MAKRYDIMAS, N. PLACHOURAS, B. THILAGANATHAN and K. H. NICOLAIDES 239

A Quantitative Estimation of the Effect of Prenatal Diagnosis in Dizygotic Twin Pregnancies in Women of Advanced Maternal Age: H. BRANDENBURG, J. H. P. VAN DER MEULEN, M. G. J. JAHODA, J. W. WLADIMIROFF, M. NIERMEIJER and J. D. F. HABBEMA 243

Prenatal Diagnosis and Dysmorphic Findings in Mosaic Trisomy 16: A. GARBER, D. CARLSON, R. SCHRECK, N. FISCHEL-GHODSIAN, W-T. HSU, S. OEZTAS, S. PEPKOWITZ and J. M. GRAHAM JR 257

The Predicitive Value of Cytogenetic Diagnosis After CVS Based on 4860 Cases with Both Direct and Culture Methods: M. C. PITTALIS, L. DALPRA, F. TORRICELLI, N. RIZZO, G. NOCERA, E. CARIATI, L. SANTARINI, M. G. TIBILETTI, S. AGOSTI, L. BOVICELLI and A. FORABOSCO 267

Comparison of First-trimester Transvaginal Amniocentesis with Chorionic Villus Sampling and Mid-trimester Amniocentesis: E. SHALEV, E. WEINER, N. YANAI, Y. SHNEUR and H. COHEN 279

Prenatal Diagnosis of X-linked Retinitis Pigmentosa (RP) in Five Pregnancies at Risk: M. P. IAMPIERI, R. MINGARELLI, E. LE GUERN, G. NOVELLI and B. DALLAPICCOLA 285

The Early Prenatal Sonographic Diagnosis of Renal Agenesis: Techniques and Possible Pitfalls: M. BRONSHTEIN, A. AMIT, R. ACHIRON, I. NOY and Z. BLUMENFELD 291

Prenatal Echocardiographic Diagnosis of Right Atrial Isomerism: V. COLLORIDI, F. PIZZUTO, F. VENTRIGLIA, A. GIANCOTTI, A PACHI and P. GALLO 299

Short Communications

Hepatic Calcifications in a Fetus with Trisomy 9 that Underwent Cordocentesis: D. SATGE, G. GASSER, A. GENEIX, P. MALET and C. STOLL 303

Prenatal Diagnosis of Twin Zygosity by DNA 'FingerPrint' Analysis: Z. APPELMAN, M. MANOR, N. MAGAL, B. CASPI, M. SHOHAT and I. BLICKSTEIN 307

A Rare Case of Cytogenetic Discrepancy Between Extraembryonic and Fetal Tissue: F. TORRICELLI, E. LISI, L. BRIZZI, M. MARIANI, R. NANNINI and E. CARIATI 310

Simpson-Golabi-Behmel Syndrome: Disproportionate Fetal Overgrowth and Elevated Maternal Serum Alpha-Fetoprotein: R. M. HUGHES-BENZIE, J. L. TOLMIE, M. MCNAY and A. PATRICK 313

Maternal Serum Free β-Human Chorionic Gonadotrophin Levels in Twin Pregnancies: Implications for Screening for Down's Syndrome: N. J. WALD and J. W. DENSEM 319

Letters to the Editor

Maternal Serum Screening for Down's Syndrome Taking Account of the Result in a Previous Pregnancy: S. HOLDING and H. CUCKLE 321

Reply to Donna Wallerstein: U. THIES 322

Euchromatic Heteromorphism or Duplication without Phenotypic Effect?: J. C. K. BARBER 323

Announcements 325

ISSUE NUMBER 5, MAY 1994

Original Papers

Chorionic Villus Sampling Utilization Following Reports of a Possible Association with Fetal Limb Defects: D. M. CUTILLO, E. A. HAMMOND, S. L REESER, M. A. KERSHNER, B. LUKIN, L. GODMILOW and A. E. DONNENFELD 327

Prenatal Diagnosis of Glutaryl-CoA Dehydrogenase Deficiency: Experience Using First-trimester Chorionic Villus Sampling: E. CHRISTENSEN 333

Variation in Prenatal Cytogenetic Diagnosis: Policies in 13 European Countries, 1989–1991: M. C. CORNEL for the EUROCAT Working Group 337

Confined Placental Mosaicism, IUGR, and Adverse Pregnancy Outcome: A Controlled Retrospective UK Collaborative Survey: J. WOLSTENHOLME, D. E. ROONEY and E. V. DAVISON 345

Cytogenetic Analysis of Chorionic Villi for Prenatal Diagnosis: An ACC Collaborative Study of UK Data: Association of Clinical Cytogeneticists Working Party on Chorionic Villi in Prenatal Diagnosis 363

Examination of Fetuses After Induced Abortion for Fetal Abnormality—A Follow-up Study: A. MEDEIRA, A. NORMAN, J. HASLAM, J. CLAYTON-SMITH and D. DONNAI 381

Maternal Serum Inhibin Levels in Second-trimester Down's Syndrome Pregnancies: H. S. CUCKLE, S. HOLDING and R. JONES 387

Second-trimester Maternal Serum Screening Using Alpha-Fetoprotein, Human Chorionic Gonadotrophin, and Unconjugated Oestriol: Experience of a Regional Programme: S. F. GOODBURN, J. R. W. YATES, P. R. RAGGATT, C. CARR, M. E. FERGUSON-SMITH, A. J. KERSHAW, P. J. D. MILTON and M. A. FERGUSON-SMITH 391

Short Communication

Multiple Marker Screen Positivity in the Presence of Hydrops Fetalis: S. KNOWLES and P. FLETT 403

Prenatal Diagnosis of Focal Nodular Hyperplasia of the Liver: B. M. PETRIKOVSKY, H. L. COHEN, Ph. SCIMECA and E. BELLUCCI 406

Terminal Deletion of Xp in a Dysmorphic Anencephalic Fetus: A. PLAJA, T. VENDRELL, E. SARRET, N. TORAN and C. MEDIANO 410

Letters to the Editor

Maternal Serum Screening and Triploidy: D. VALERIO 413

Prenatal Diagnosis of Severe Fetal Toxoplasmosis as a Result of Toxoplasmic Reactivation in an HIV-1 Seropositive Woman: P. MARTY, A. BONGAIN, A. RAHAL, P. THULLIEZ, D. WASFI, J. C. LAMBERT, Y. LE FICHOUX AND J. Y. GILLET 414

Re: Diagnostic Potential of Fetal Renal Biopsy: M. E. FURNESS 415

Prenatal Diagnosis of X-Linked Hydrocephalus: L. STRAIN, D. J. H. BROCK and D. T. BONTHRON 415

Prenatal Diagnosis of Combined Methylmalonic Aciduria and Homocystinuria (Cobalamin CblC or CblD Mutant): B. CHADEFAUX-VEKEMANS, M. O. ROLLAND, S. LYONNET, D. RABIER, P. DIVRY and P. KAMOUN 417

Announcement 419

ISSUE NUMBER 6, JUNE 1994

Original Papers

Congenital Malformations Subsequent to Chorionic Villus Sampling: Outcome Analysis of 1048 Consecutive Procedures: R. K. SILVER, S. N. MacGREGOR, L. H. MUHLBACH, T. A. KNUTEL and M. P. KAMBICH 421

Humerus and Femur Length in Fetuses with Down Syndrome: R. BIAGIOTTI, E. PERITI and E. CARIATI 429

Prenatal Diagnosis of Pyruvate Dehydrogenase E1α Subunit Deficiency: R. M. BROWN and G. K. BROWN 435

Prenatal Diagnosis of Tyrosinase-Negative Oculocutaneous Albinism by an Electron Microscopic DOPA Reaction Test of Fetal Skin: H. SHIMIZU, A. ISHIKO, A. KIKUCHI, M. AKIYAMA, K. SUZUMORI and T. NISHIKAWA 443

Trisomy 8 Mosaicism in Chorionic Villus Sampling: Case Report and Counselling Issues: J. KLEIN, J. M. GRAHAM Jr., L. D. PLATT and R. SCHRECK 451

Screening Maternal Serum Alpha-Fetoprotein Levels and Human Parvovirus Antibodies: D. R. JOHNSON, R. A. FISHER, J. J. HELWICK, D. L. MURRAY, M. J. PATTERSON and F. P. DOWNES 455

First-Trimester Prenatal Diagnosis of Spinal Muscular Atrophy Using Microsatellite Markers: S. LO CICERO, F. CAPON, S. MELCHIONDA, M. GENNARELLI, G. NOVELLI and B. DALLAPICCOLA 459

Fetal Nuchal Oedema and Antenatal Diagnosis of Trisomy 10: S. A. FARRELL, A. SUE-CHUE-LAM, M. MISKIN and Y.-S. FAN 463

Prenatal Diagnosis of Fragile X Syndrome: Management of the Male Fetus with a Premutation: L. STRAIN, M. E. M. PORTEOUS, C. M. GOSDEN, P. M. ELLIS, J. P. NEILSON and D. T. BONTHRON 469

Prenatal Diagnosis of Glutathione Synthase Deficiency: N. J. MANNING, N. P. DAVIES, S.E. OLPIN, K. H. CARPENTER, M. F. SMITH, R. J. POLLITT, S. L. B. DUNCAN, A. LARSSON and B. CARLSSON 475

Comparison of Chorionic Villus Sampling and Amniocentesis: Current Status of Prenatal Diagnosis in Japan: K. SUZUMORI, S. OKADA, R. ADACHI, T. IIDA, T. SUMI, M. TANEMURA and Y. YAGAMI 479

Prediction of an Abnormal Karyotype in Fetuses with Omphalocele: M. DE VECIANA, C. A. MAJOR and M. PORTO 487

A New RFLP Marker, SP282 at the *btk* Locus for Genetic Analysis in X-linked Agammaglobulinaemia Families: S.-P. KWAN, A. P. WALKER, T. HAGEMANN, S. GUPTA, B. VAYUVEGULA and H. D. OCHS 493

Short Communication

Early Amniocentesis — A Cytogenetic Evaluation of Over 1500 Cases: B. EIBEN, R. GOEBEL, S. HANSEN and W. HAMMANS 497

Prenatal Diagnosis of Femur-Fibula-Ulna Complex by Ultrasonography in a Male Fetus at 24 Weeks of Gestation: G. CAPECE, A. FASOLINO, M. DELLA MONICA, F. LONARDO, G. SCARANO and G. NERI 502

Prenatal Diagnosis and Post-mortem Study of a Fetus with Mosaic Trisomy 14 Due to a dic(14) (p11): I. LAMBERT, J. KEMP, J. JACKSON, H. JOYCE, S. MANN, A. KAN and A. SMITH 507

Letter to the Editor

Prenatal Diagnosis of Roberts Syndrome: A. GRUBER, D. RABINERSON, B. KAPLAN and Y. OVADIA 511

ISSUE NUMBER 7, JULY 1994

Original Papers

Allele Frequencies and Molecular Diagnosis in Haemophilia A and B Patients from Russia and from some Asian Republics of the Former U.S.S.R.: M. ASEEV, V. SURIN, K. BABOEV, N. GORNOSTAEVA, T. KUZNETZOVA, T. KASCHEEVA, T. IVASCHENKO, G. SOLOVYEV, A. MIKHAILOV, V. LEBEDEV, L. PAPAIAN, T. ANDREEVA, D. M. AMOASHYI, V. VAKHARLOVSKY and V. S. BARANOV 513

Prenatal Ultrasonographic Diagnosis of Fetal Cerebral Ventriculitis Associated with Asymptomatic Maternal Cytomegalovirus Infection: R. ACHIRON, O. PINHAS-HAMIEL, S. LIPITZ, Z. HEIMAN, B. REICHMAN and S. MASHIACH 523

Incremental Cost-effectiveness of Incorporating Oestriol Evaluation in Down Syndrome Screening Programmes: T. G. GANIATS, A. L. HALVERSON and M. H. BOGART 527

Down's Syndrome Screening in Multiple Pregnancies Using Alpha-fetoprotein and Free Beta hCG: K. SPENCER, R. SALONEN and F. MULLER 537

Maternal Age-specific Risks for Trisomies at 9–14 Weeks' Gestation: R. J. M. SNIJDERS, W. HOLZGREVE, H. CUCKLE and K. H. NICOLAIDES 543

Early Invasive Prenatal Diagnosis in HBsAg-Positive Women: P. M. GROSHEIDE, H. W. P. QUARTERO, S. W. SCHALM, R. A. HEIJTINK and G. C. M. L. CHRISTIAENS 553

True Trisomy 15 Mosaicism, Detected by Amniocentesis at 12 Weeks of Gestation and Fetal Echocardiography: K. SUNDBERG, V. BROCKS, J. R. JACOBSON and B. BECK 559

Maternal Serum Markers in Second-trimester Oligohydramnios: F. J. LOS, A. M. HAGENAARS, T. E. COHEN-OVERBEEK and H. W. P. QUARTERO 565

Discrepant Karyotypes After Second- and Third-trimester Combined Placentacentesis/Amniocentesis: D. CASPARI, I. BARTELS, R. RAUSKOLB, G. PRANGE, R. OSMERS and B. EIBEN 569

Prenatal Diagnosis of Sjögren-Larsson Syndrome Using Enzymatic Methods: W. B. RIZZO, D. A. CRAFT, T. L. KELSON, J.-P. BONNEFONT, J.-M. SAUDUBRAY, J. D. SCHULMAN, S. H. BLACK, K. TABSH, M. DiROCCO and R. J. McKINLAY GARDNER . 577

Characterization of Marker Chromosomes By Microdissection and Fluorescence *in Situ* Hybridization: M. THANGAVELU, E. PERGAMENT, R. ESPINOSA III and S. K. BOHLANDER 583

Confined Placental Mosaicism in CVS and Pregnancy Outcomes: R. ROLAND, L. LYNCH, G. BERKOWITZ and R. ZINBERG 589

Complex Karyotypic Mosaicism as a Result of Non-disjunction and Subsequent Centromere Fission: G. PRABHAKAR, S. SCHWARTZ, E. WATERS, C. PUNZALAN and M. M. COHEN 595

Congenital Nephrosis in Low-risk Pregnancies: A. GHIDINI, M. ALVAREZ, G. SILVERBERG, E. AINBENDER and C. J. LOCKWOOD 599

Repeat Maternal Serum Testing in Multiple Marker Down's Syndrome Screening Programmes: H. CUCKLE, J. DENSEM and N. WALD 603

Adverse Perinatal Outcome in Patients Screen-positive for Neural Tube Defects and Fetal Down Syndrome: S. J. GROSS, O. P. PHILLIPS, L. P. SHULMAN, N. L. BRIGHT, J. S. DUNGAN, J. L. SIMPSON and S. ELIAS 609

Antenatal Detection of Neural Tube Defects: Comparison of Biochemical and Immunofluorescence Methods: J. E. BELL, L. BARRON and G. RAAB 615

Antenatal Diagnosis of a Congenital Nasolacrimal Duct Cyst by Ultrasonography: A Case Report: C. M. ALPER, K. H. CHAN, L. M. HILL and P. CHENEVEY 623

Cardiac Blood Flow Studies in Fetuses with Haemoglobin Bart's Disease: A. GHOSH, M. TAN, M. P. LEUNG, L. M. FUNG and H. K. MA 627

Short Communications

Do Radical Differences Exist in Second-trimester Maternal hCG Levels? A Study of 23 369 Patients: F. MULLER, L. BUSSIÈRES, M.-C. PÉLISSIER, J.-F. OURY, C. BOUÉ, S. UZAN and A. BOUÉ 633

Prenatal Diagnosis of Congenital Cystic Adenomatoid Malformation of the Lung by Fetal Lung Biopsy: J. MUROTSUKI, S. UEHARA, K. OKAMURA, A. YAJIMA and K. MURAKAMI 637

Monosomy 8q: Prenatal Diagnosis and Autopsy Findings: I. ARIEL, E. ANTEBY, D. SOFFER, M. TOBIAS and S. YAGEL 640

The 48.XXYY Syndrome: A Case Detected by Maternal Serum Alpha-fetoprotein Screening: R. H. NYBERG, R. KARHU, R. KARIKOSKI and K. O. J. SIMOLA 644

Letters to the Editor

Down Syndrome and Fetal Ventriculomegaly: S. A. FARRELL 647

Why Karyotype Cystic Hygroma Cells?: M.-R. VERSCHRAEGEN-SPAE and A. DE PAEPE 649

Neonatal Convulsions Following CVS: G. MASON, R. LILFORD, R. MUELLER and G. LINTON 650

Pitfall: Amniocentesis Fails to Detect Mosaic Trisomy 8 in a Male Newborn: M. SCHNEIDER, U. KLEIN-VOGLER, J. TOMIUK, M. SCHLIEPHACKE, M. LEIPOLDT and H. ENDERS 651

ISSUE NUMBER 8, AUGUST 1994

Original Papers

Prenatal Sonographic Detection of Intra-abdominal Extralobar Pulmonary Sequestration: Report of Three Cases and Literature Review: J. WHITE, Y.-F. CHAN, S. NEUBERGER and T. WILSON 653

Results from a Reference Laboratory for Prenatal Diagnosis of Sickle Cell Disorders in Cuba: H. GRANDA, S. GISPERT, G. MARTINEZ, M. GOMEZ, R. FERREIRA, T. COLLAZO, C. MAGARIÑO and L. HEREDERO 659

Isolated Choroid Plexus Cysts—The Need for Routine Offer of Karyotyping: S.A. WALKINSHAW, D. PILLING and A. SPRIGGS 663

Body Stalk Anomaly Associated with Maternal Cocaine Abuse: J. M. MARTINEZ, A. FORTUNY, C. COMAS, B. PUERTO, A. BORRELL, M. PALACIO and O. COLL 669

Prenatal Diagnosis and Long Survival of Fryns' Syndrome: E. C. GADOW, S. LIPPOLD, E. SERAFIN, L. J. SALGADO, C. GARCIA and L. PRUDENT 673

Cystic Adenomatoid Malformation of the Lung: Prenatal Diagnosis and Outcome: J. G. THORPE-BEESTON and K. H. NICOLAIDES 677

Karyotype Abnormalities in Fetuses Diagnosed as Abnormal on Ultrasound Before 20 Weeks' Gestational Age: J. HALLIDAY, J. LUMLEY and A. BANKIER 689

Value of Maternal Serum Unconjugated Oestriol Measurement in Prenatal Screening for Down's Syndrome: N. J. WALD, A. KENNARD, H. C. WATT and S. SMITH ... 699

Four-marker Serum Screening for Down's Syndrome: N. J. WALD, J. W. DENSEM, D. SMITH and G. G. KLEE 707

Maternal Serum Free α-Human Chorionic Gonadotrophin Levels in Twin Pregnancies: Implications for Screening for Down's Syndrome: N. J. WALD and J. W. DENSEM 717

Prenatally Diagnosed Non-immune Hydrops Caused by Congenital Transient Leukaemia: A. E. DONNENFELD, S. C. SCOTT, M. HENSELDER-KIMMEL and C. D. DAMPIER 721

Gorlin Syndrome (Naevoid Basal Cell Carcinoma Syndrome): Prenatal Detection in a Fetus with Macrocephaly and Ventriculomegaly: W. A. HOGGE, C. BLANK, L. B. ROOCHVARG, J. S. HOOGE, E. A. WULFSBERG and L. J. RAFFEL 725

The Imprecision in Rates of Down Syndrome by 1-year Maternal Age Intervals: A Critical Analysis of Rates Used in Biochemical Screening: C.A. HECHT and E. B. HOOK 729

Fetal Ear Measurements in the Prenatal Detection of Trisomy 21: P. GILL, J. VANHOOK, J. FITZSIMMONS, J. PASCOE-MASON AND A. FANTEL 739

Prenatal Diagnosis of Klippel-Trénaunay-Weber Syndrome by Ultrasound: J. YANKOWITZ, D. D. SLAGEL and R. WILLIAMSON 745

Human Maternal Uniparental Disomy for Chromosome 16 and Fetal Development: J. VAUGHAN, Z. ALI, S. BOWER, P. BENNETT, T. CHARD and G. MOORE 751

Short Communications

Late-onset Isolated Cystic Hygroma: the Obstetrical Significance, Management and Outcome: I. GOLDSTEIN, P. JAKOBI, G. SHOSHANY, S. FILMER, I. ITSKOWIZ and B. MAOR 757

Fetal Hypothyroidism as a Complication of Amiodarone Treatment for Persistent Fetal Supraventricular Tachycardia: L. DE CATTE, D. DE WOLF, J. SMITZ, A. BOUGATEF, J. DE SCHEPPER and W. FOULON 762

Prenatal Sonographic Appearance of Foramina Parietalia Permagna: A. SALAMANCA, F. GONZALEZ-GOMEZ, M. C. PADILLA, M. A. MOTOS, F. ZORRILLA and R. M. SABATEL 766

Prenatal Ultrasound Diagnosis of Rhizomelic Chondrodysplasia Punctata in a Primigravida: S. H. SASTROWIJOTO, K. VANDENBERGHE, P. MOERMAN, J. M. LAUWERYNS and J.-P. FRYNS 770

ISSUE NUMBER 9, SEPTEMBER 1994

Original Papers

Prenatal Diagnosis and Carrier Detection in Mucopolysaccharidosis Type II by Mutation Analysis. A 47,XXY Male Heterozygous for a Missense Point Mutation: S.BUNGE, C. STEGLICH, P. LORENZ, M. BECK, S. XU, J. J. HOPWOOD and A. GAL 777

Evaluation of Prenatal Diagnosis of Limb Reduction Defects by a Registry of Congenital Anomalies: C. STOLL, Y. ALEMBIK, B. DOTT and M. P. ROTH 781

Tetrasomy 12p (Pallister-Killian Syndrome): Ultrasound Indicators and Confirmation by Interphase FISH: R. D. WILSON, K. HARRISON, L. A. CLARKE and S. L. YONG . 787

The Association between Alpha-fetoprotein and β-hCG Levels Prior to and Following Chorionic Villus Sampling in Cases that Spontaneously Miscarried: G. BARKAI, B. REICHMAN, L. RIES, A. LUSKY, S. LIPITZ and B. GOLDMAN 793

Umbilical Artery Velocity Waveforms Before And After Chorionic Villus Sampling: R. M. IBBA, G. MONNI, G. OLLA and A. CAO 799

Transplacental Amniocentesis: Is it Really a Higher-risk Procedure?: C. GIORLANDINO, L. MOBILI, E. BILANCIONI, P. D'ALESSIO, O. CARCIOPPOLO, P. GENTILI and A. VIZZONE 803

The Value of Sonographic Diagnosis of Fetal Malformations: Different Results between Indication-based and Screening-based Investigations: G. BERNASCHEK, I. STUEMPFLEN and J. DEUTINGER 807

Pitfalls in the Prenatal Diagnosis of Peroxisomal β-Oxidation Defects by Chorionic Villus Sampling: W. F. CAREY, A. POULOS, P. SHARP, P. V. NELSON, E. F. ROBERTSON, J. L. HUGHES and A. GILL 813

Feto-amniotic Shunting—Report of the Experience of Four European Centres: G. BERNASCHEK, J. DEUTINGER, M. HANSMANN, R. BALD, W. HOLZGREVE and R. BOLLMANN 821

Maternal Serum Free α- and Free β-Human Chorionic Gonadotrophin in Pregnancies with Insulin-dependent Diabetes Mellitus: Implications for Screening for Down's Syndrome: N. J. WALD, J. W. DENSEM, R. CHENG and S. COLLISHAW 835

Syndromes Identified in Fetuses with Prenatally Diagnosed Cephaloceles: S. J. WINNIGER and A. E. DONNENFELD 839

Second-trimester Echogenic Small Bowel: An Increased Risk for Adverse Perinatal Outcome: L. M. HILL, J. FRIES, J. HECKER and P. GRZYBEK..... 845

Experience with 500 Prenatal Diagnoses of Sickle Cell Diseases: The Effect of Gestational Age on Affected Pregnancy Outcome: X. WANG, C. SEAMAN, M. PAIK, T. CHEN, A. BANK and S. PIOMELLI 851

Reasons for Women's Non-uptake of Amniocentesis: C. JULIAN-REYNIER, G. MACQUART-MOULIN, J.-P. MOATTI, Y. AURRAN, F. CHABAL and S. AYMÉ..... 859

Short Communications

Prenatal Sonographic Diagnosis of a Third Ventricle Choroid Plexus Papilloma: A. M. ADRA, A. A. MEJIDES, F. A. SALMAN, H. J. LANDY and A. W. HELFGOTT..... 865

Minute Chromatin Structures in Cells of Amniotic Fluid—An Interpretive Dilemma: H. WANG, J. L. BELLINGER, K. BRIERLEY, L. E. DAWSON, C. L. GOLDSMITH and A. G. W. HUNTER 868

Fetal Renal Vein Thrombosis, Hydrops Fetalis and Maternal Lupus Anticoagulant. A Case Report: M. L. HAGE, R. LIU, D. G. MARCHESCHI, J. D. BOWIE, N. B. ALLEN and B. G. MACIK 873

Prenatal Diagnosis of Trisomy 12 Mosaicism; Physical and Developmental Follow-up: J. M. MECK, C. KOZMA, J.-G. TCHABO, J. C. KING, S. LENCKI and T. L. PINCKERT 878

***In Utero* Development of a Mediastinal Teratoma: A Second-trimester Event:** M. KENT FROBERG, R. E. BROWN, J. MAYLOCK and E. POLING 884

Prenatal Diagnosis of Congenital Gastric Outlet Obstruction: A. WEISSMAN, R. ACHIRON, J. KUINT, S. LIPITZ, S. MASHIACH and I. AVIGAD 888

Prenatal Evaluation and *In Utero* Platelet Transfusion for Thrombocytopenia Absent Radii Syndrome: M. WEINBLATT, B. PETRIKOVSKY, M. BIALER, J. KOCHEN and R. HARPER 892

Letters to the Editor

Amniotic Fluid Leakage and Miscarriages after TA-CVS: R. SAURA, L. TAINE, Z.Q. WEN, W. TRAORE and J. HOROVITZ 897

Comparison of Two Down Syndrome Screening Strategies for Women Age 35 and Older: G. E. PALOMAKI and J. E. HADDOW..... 898

Pregnancy-associated Plasma Protein-A (PAAP-A), a First-trimester Screening Test for Down Syndrome and Other Chromosomal Anomalies: B. BRAMBATI, M. C. M. MACINTOSH, K. SHRIMANKER, T. CHARD and J. G. GRUDZINSKAS 899

Announcement 901

ISSUE NUMBER 10, OCTOBER 1994

Prenatal Diagnosis of Congenital Human Cytomegalovirus Infection: U. NICOLINI, A. KUSTERMANN, B. TASSIS, R. FOGLIANI, A. GALIMBERTI, E. PERCIVALLE, M. G. REVELLO and G. GERNA 903

Tumour-associated Antigens in Maternal and Fetal Blood: P. HOHLFELD, T.-T. DANG, K. NAHOUL, F. DAFFOS and F. FORESTIER..... 907

Risk Assessment of Amniocentesis Between 11 and 15 Weeks: Comparison to Later Amniocentesis Controls: B. F. CRANDALL, P. KULCH and K. TABSH 913

Prenatal Diagnosis of Xeroderma Pigmentosum and Cockayne Syndrome: J. E. CLEAVER, J. P. G. VOLPE, W. C. CHARLES and G. H. THOMAS 921

Identification of a Novel β^0-Thalassaemia Mutation in a Greek Family and Subsequent Prenatal Diagnosis: J. S. WAYE, B. ENG, N. G. OLIVIERI and D. K. CHUI .. 929

Postnatal Confirmation of Prenatally Diagnosed Trisomy 16 Mosaicism in Two Phenotypically Abnormal Liverborns: B. A. PLECHTER, M. M. SANZ, J. S. SCHLESSEL, S. KUNAPORN, M. MCKENNA, M. G. BIALER, M. L. ALONSO, A.-L. ZASLAV, W. T. BROWN and J. H. RAY 933

The Causes and Natural History of Fetal Ascites: C. ZELOP and B.R. BENACERRAF 941

Second-trimester Diurnal Variation of Maternal Serum Alpha-fetoprotein, Human Chorionic Gonadotropin, and Unconjugated Oestriol: Is it Present and Does it Affect Prediction of a Patient's Risk for Fetal Down Syndrome?: N. C. ROSE, J. A. CANICK, G. J. KNIGHT, A. PULKKINEN, M. B. TUMBER, M. T. MENNUTI and G. E. PALOMAKI 947

Urinary β-Core Human Chorionic Gonadotrophin: A New Approach to Down's Syndrome Screening: H. S. CUCKLE, R. K. ILES and T. CHARD 953

Prenatal Diagnosis by Minisatellite Analysis in Italian Families with Phenylketonuria: V. ROMANO, I. DIANZANI, A. PONZONE, E. ZAMMARCHI, R. EISENSMITH, N. CERATTO, P. BOSCO and A. INDELICATO 959

First-trimester Maternal Serum Alpha-Fetoprotein as a Marker for Fetal Chromosomal Disorders: J. M. M. VAN LITH for the Dutch Working Party on Prenatal Diagnosis 963

Amniotic Fluid Digestive Enzymes: Diagnostic Value in Fetal Gastrointestinal Obstructions: F. MULLER, M. DOMMERGUES, Y. VILLE, F. LEWIN, N. DEVALEZ-MORICHON, C. NIHOUL-FEKETE, F. BARGY, Y. DUMEZ and A BOUE 973

Hyperechoic Fetal Bowel: The Perinatal Consequences: R. BAHADO-SINGH, R. MOROTTI, J. A. COPEL and M. J. MAHONEY 981

Short Communications

Prenatal Diagnosis of the Klippel-Trenaunay-Weber Syndrome: R. J. JORGENSON, B. DARBY, R. PATTERSON and K. J. TRIMMER 989

Intrauterine Intervention in a Case of Recurrent Meconium Peritonitis: M.-K. SHYU, C.-D. CHEN, F.-J. HSIEH, K.-I. T. YAU, G.-J. LIN and M.-L. LEE 993

Prenetal Diagnosis of Bartter Syndrome: H. SHALEV, M. OHALY, I. MEIZNER and R. CARMI 996

Improvement of Prenatal Diagnosis of Wilson Disease Using Microsatellite Markers: G. LOUDIANOS, A. L. FIGUS, A. LOI, A. ANGIUS, V. DESSI, M. DEIANA, S. DE VIRGILIIS, G. MONNI, A. CAO and M. PIRASTU 999

Successful Outcome of Pregnancy After Treatment of Maternal Anti-Ro (SSA) Antibodies with Immunosuppressive Therapy and Plasmapheresis: J. N. VAN DER LEIJ, G. H. A. VISSER, M. Th. E. BINK-BOELKENS, J. F. MEILOF and C. G. M. KALLENBERG 1003

Announcements i

ISSUE NUMBER 11, NOVEMBER 1994

Original Papers

Exclusion of Chromosomal Mosaicism in Amniotic Fluid Cultures: Determination of Number of Colonies Needed for Accurate Analysis: T. FEATHERSTONE, S. W. CHEUNG, E. SPITZNAGEL and D. PEAKMAN 1009

Swedish Survey On Extra Structurally Abnormal Chromosomes in 39 105 Consecutive Prenatal Diagnoses: Prevalence and Characterization by Fluorescence *in Situ* Hybridization: E. BLENNOW, T.-H. BUI, U. KRISTOFFERSSON, M. VUJIC, G. ANNERÉN, E. HOLMBERG and M. NORDENSKJÖLD 1019

Prenatal Diagnosis of the Derivative Chromosome 22 Associated with Cat Eye Syndrome by Fluorescence *In Situ* Hybridization: S. L. REESER, A. E. DONNENFELD, R. C. MILLER, B. S. SELLINGER, B. S. EMANUEL and D. A. DRISCOLL 1029

Prenatal Ultrasonic Diagnosis of Obstructive Bowel Disease: A Retrospective Analysis: R. HEYDANUS, M. C. SPAARGAREN and J. W. WLADIMIROFF 1035

Serum PAPP-A and Free β-hCG are First-Trimester Screening Markers for Down Syndrome: B. BRAMBATI, L. TULUI, I. BONACCHI, K. SHRIMANKER, Y. SUZUKI and J. G. GRUDZINSKAS 1043

Prenatal Detection of Trisomy 21 in Uncultured Amniocytes by Fluorescence *In Situ* Hybridization: A Prospective Study: D. H. SPATHAS, A. DIVANE, G. M. MANIATIS, M. E. FERGUSON-SMITH and M. A. FERGUSON-SMITH 1049

Accuracy of Amniotic Fluid Testing Before 21 Weeks' Gestation in Prenatal Diagnosis of Congenital Cytomegalovirus Infection: C. DONNER, C. LIESNARD, F. BRANCART and F. RODESCH 1055

Rapid Prenatal Diagnosis of Aneuploidy from Uncultured Amniotic Fluid Cells Using Five-Colour Fluorescence *In Situ* Hybridization: A. DIVANE, N. P. CARTER, D. H. SPATHAS and M. A. FERGUSON-SMITH 1061

Short Communications

Diagnosis of Bloom's Syndrome by Sister Chromatid Exchange Evaluation in Chorionic Villus Cultures: R. T. HOWELL and T. DAVIES 1071

Prenatal Activity of a Fetus with Early-Onset, Severe Spinal Muscular Atrophy: P. KIRKINEN, M. RYYNÄNEN, P. HARING, H. TORKKELI, L. PÄÄKKÖNEN and A. MARTIKAINEN 1074

The Amplification Refractory Mutation System (ARMS): A Rapid and Direct Prenatal Diagnostic Technique for β-Thalassaemia in Singapore: J. A. M. A. TAN, J. S. H. TAY, L. I. LIN, S. K. Y. KHAM, J. N. CHIA, T. M. CHIN, NORKAMAR BT. ADB. AZIZ and H. B. WONG 1077

Measurement of Fetal Urine Production in Mild Infantile Polycystic Kidney Disease—A Case Report: H. TSUDA, M. MATSUMOTO, M. IMANAKA, S. OGITA 1083

MSAFP Levels and Oesophageal Atresia: B. N. CHODIRKER, A. E. CHUDLEY, K. M. MACDONALD, C. R. HARMAN and J. A. EVANS 1086

Prenatal Diagnosis of Renal Agenesis in a Twin Gestation: J. A. KULLER, C. C. COULSON, M. C. McCOY, G. C. ALTMAN, J. M. THORP Jr. and V. L. KATZ 1090

Letters to the Editor

Maternal Serum Free Beta hCG Screening: Results of Studies Including 480 Cases of Down Syndrome—Problems with this Approach: A. S. WIERZBICKI 1093

Maternal Serum Free Beta hCG Screening: R. ZIMMERMANN 1094

Authors' Reply to Wierzbicki and Zimmermann: J. N. MACRI and K. SPENCER 1094

The 18ph+ Chromosome Heteromorphism: L. ZELANTE, A. NOTARANGELO and B. DALLAPICCOLA 1096

Response to Zelante *et al.*: M. C. PITTALIS, L. SANTARINI and L. BOVICELLI 1097

Prenatal Diagnosis of Partial 2p Trisomy by *'de novo'* Duplication 2p (13.1→21). Confirmation by FISH: J. P. S. SIFFROI, D. MOLINA-GOMEZ, F. VIGUIE, C. NESSMANN and J. P. DADOUNE 1097

On the Inheritance of Harlequin Ichthyosis: A. BOTTANI 1099

ISSUE NUMBER 12, DECEMBER 1994

Intrauterine Growth Retardation and Fetal Transverse Cerebellar Diameter: R. J. M. SNIJDERS, R. A. B. DE COURCY-WHEELER and K. H. NICOLAIDES 1101

Does Chorionic Villus Sampling Compromise Fetal Umbilical Blood Flow?: J. U. HIBBARD, G. L. LOY and M. C. HIBBARD 1107

A Simple, Rapid, and Highly Informative PCR-based Procedure for Prenatal Diagnosis and Carrier Screening of Phenylketonuria: R. C. EISENSMITH, A. A. GOLTSOV and S. L. C. WOO 1113

Fetal Choroid Plexus Cysts and Trisomy 18: Assessment of Risk Based on Ultrasound Findings and Maternal Age: R. J. M. SNIJDERS, L. SHAWA and K. H. NICOLAIDES 1119

Enrichment of Fetal Cells From Maternal Blood by High Gradient Magnetic Cell Sorting (Double MACS) For PCR-Based Genetic Analysis: J. BÜSCH, P. HUBER, E. PFLUGER, St. MILTENYI, J. HOLTZ and A. RADBRUCH 1129

Flow Cytometric Method to Measure Urea-resistant Alkaline Phosphatase from Blood Neutrophils for Use in the Prenatal Screening of Down's Syndrome: Y. P. AGRAWAL, P. YLIKANGAS, M. T. PARVIAINEN and I. M. PENTTILÄ 1141

Short Communications

Prenatal Diagnosis of Tuberous Sclerosis. Use of Magnetic Resonance Imaging and Its Implications for Prognosis: H. WERNER, Jr., V. MIRLESSE, F. JACQUEMARD, P. SONIGO, A. L. DELEZOIDE, M. GONZALES, P. BRUNELLE, L. FERMONT and F. DAFFOS 1151

Early Detection of Abdominal Pregnancy by Maternal Serum AFP+ Screening: A. T. BOMBARD, S. NAKAGAWA, C. D. RUNOWICZ, B. L. COHEN, M. S. MIKHAIL and H. M. NITOWSKY 1155

Gene Therapy for Cystic Fibrosis: Will it Affect the Uptake of Prenatal Carrier Screening?: M. E. MENNIE, M. E. COMPTON, W. A. LISTON and D. J. H. BROCK ... 1158

Chromosome Preparation of Both Direct and Long-term Cultures from the Same Fragment of Chorionic Villi: J. M. DAWSON and M. J. GRIFFITHS 1163

Prenatal Detection of Congenital Hypospadias in the Wolf-Hirschhorn (4p–) Syndrome: F. VIÑALS, W. SEPULVEDA and E. SELMAN 1166

Sonographic Presentation of Hirschsprungs's Disease. A Case of an Entirely Aganglionic Colon and Ileum: S. ELIYAHU, N. YANAI, O. BLONDHEIM, D. REICH, L. SIPLOVICH and E. SHALEV 1170

Letters to the Editor

Prenatal Diagnosis of Exencephaly Associated with High Levels of Maternal Serum Human Chorionic Gonadotropin and Urinary β-core Fragment of hCG/ Creatinine Ratio: M. HAYASHI, I. HIROYUKI, T. SODEMOTO and T. SHINAGAWA ... 1173

Fetal Detection of DUP 9p11–12: G. CALABRESE, L. STUPPIA, P. GUANCIALI-FRANCHI, R. MINGARELLI and G. PALKA 1174

Compound Heterozygosity for ΔF508 and F508C: a Cautionary Note on the Molecular Diagnosis of Cystic Fibrosis: R. DUFOURCQ, S. VUILLAUMIER, O. PASCAUD, C. GUIDAL, J.-F. OURY, J. ELION and E. DENAMUR 1176

ISSUE NUMBER 13, ANNUAL REVIEW ISSUE, DECEMBER 1994

Publisher's note 1179

Editorial 1181

Confined Placental Mosaicism: G. SIMONI and S. M. SIRCHIA 1185

Genomic Imprinting Related to Prenatal Diagnosis: D. K. KALOUSEK and I. BARRETT 1191

Prenatal Aneuploidy Detection in Interphase Cells by Fluorescence *In Situ* Hybridization (FISH): J. PHILIP, T. BRYNDORF and B. CHRISTENSEN 1203

Preimplantation Diagnosis: J. D. A. DELHANTY 1217

Isolating Fetal Cells in Maternal Circulation for Prenatal Diagnosis: J. L. SIMPSON and S. ELIAS 1229

Carrier Screening for Cystic Fibrosis: D. J. H. BROCK 1243

Prenatal Diagnosis in Canada—1990: a Review: L. C. STRANC, J. A. EVANS and J. L. HAMERTON 1253

Author Index i

Key Word Index v

Volume contents x